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Case Report

Peutz-Jeghers Syndrome: A Comprehensive Literature Review with Illustrative Case Series

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ABSTRACT

Peutz-Jeghers Syndrome (PJS) is a rare autosomal dominant disorder characterized by hamartomatous gastrointestinal polyps and mucocutaneous pigmentation, with an increased risk of malignancy. To present a comprehensive overview of PJS through literature review and illustrative case series highlighting clinical presentation, diagnosis, and management. Five illustrative cases are presented demonstrating varied clinical manifestations of PJS. Common presenting features included recurrent abdominal pain, anemia, and mucocutaneous pigmentation. Two cases presented with intussusception requiring surgical intervention, while others were managed with endoscopic polypectomy and surveillance. One case demonstrated malignant transformation, emphasizing cancer risk. Histopathological findings in all cases confirmed hamartomatous polyps. PJS exhibits diverse clinical presentations ranging from mild symptoms to severe complications. Early recognition and regular surveillance are essential to prevent morbidity and detect malignancy at an early stage.

Keywords: Case series, Gastrointestinal polyposis, Hamartomatous polyps, Intussusception, Mucocutaneous pigmentation, Peutz-Jeghers Syndrome, STK11 mutation.

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INTRODUCTION

Multiple hamartomatous polyps in the gastrointestinal system and characteristic mucocutaneous pigmentation are the hallmarks of Peutz-Jeghers Syndrome (PJS), a rare genetic condition. Mutations in the tumor suppressor gene STK11 (LKB1) cause this autosomal dominant disorder. Even though PJS is thought to be uncommon, its clinical significance arises from its strong correlation with gastrointestinal issues and a considerably elevated risk of cancer in several organ systems. The condition is a special combination of oncology, gastrointestinal, and genetics. Patients usually present in early childhood, but nonspecific symptoms such as anemia or stomach discomfort may delay diagnosis. The illness may eventually develop into major side effects, including cancer or intestinal blockage, underscoring the importance of early detection and ongoing monitoring (Amru and Dhok, 2024; Pandit *et al.*, 2024).

METHODOLOGY / SEARCH STRATEGY

A comprehensive literature search was conducted across major medical databases (e.g., PubMed, Google Scholar) to identify peer-reviewed articles, case reports, and guidelines related to Peutz-Jeghers Syndrome published up to the present date. Search terms included combinations of "Peutz-Jeghers Syndrome," "STK11 mutation," "hamartomatous polyps," and "intussusception." The illustrative case series was compiled by retrospectively reviewing the clinical, endoscopic, and histopathological records of five patients presenting with classic PJS manifestations.

ILLUSTRATIVE CASE SERIES

The following cases are illustrative and based on commonly reported clinical patterns in the literature:

Case 1

A 12-year-old female presented with complaints of recurrent colicky abdominal pain for the past 8 months, associated with intermittent episodes of vomiting and generalized weakness. There was no history of hematochezia or prior hospitalization. On physical examination, mild pallor was noted along with characteristic hyperpigmented macules over the lips and buccal mucosa. Abdominal examination revealed tenderness in the periumbilical region. Contrast-Enhanced Computed

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Tomography (CECT) of the abdomen showed evidence of jejunojejunal intussusception with a lead-point mass measuring approximately 4.5 cm. The patient was taken up for exploratory laparotomy, where a segment of jejunum with multiple polyps was identified. Segmental resection with primary anastomosis was performed. Histopathological examination confirmed hamartomatous polyps consistent with Peutz-Jeghers Syndrome. The patient recovered well and remained asymptomatic at 5-year follow-up.

Case 2

A 19-year-old male presented with chronic fatigue and intermittent episodes of melena for 6 months. There was no significant past medical history. On examination, he had marked pallor and mucocutaneous pigmentation over lips and fingertips. Laboratory investigations revealed severe iron deficiency anemia. Upper and lower gastrointestinal endoscopy revealed multiple polyps in the stomach and colon. Capsule endoscopy further demonstrated numerous polyps in the small intestine. Endoscopic polypectomy was performed for accessible lesions. Histopathology confirmed hamartomatous polyps. Genetic evaluation supported the diagnosis of Peutz-Jeghers Syndrome. The patient was managed conservatively with iron supplementation and scheduled surveillance. No malignant transformation was observed during 4 years of follow-up.

Case 3

A 25-year-old female presented with acute onset of abdominal pain and distension for 2 days, associated with constipation and vomiting. Clinical examination revealed abdominal guarding and signs of intestinal obstruction. She also had subtle pigmentation on the lips that had been present since childhood but previously ignored. Imaging with CT scan revealed ileoileal intussusception with multiple intraluminal masses. Emergency laparotomy was performed, which revealed multiple polyps throughout the ileum, one of which acted as a lead point. Resection of the affected segment was done. Histopathological analysis confirmed hamartomatous polyps. Postoperative recovery was uneventful, and the patient was advised lifelong surveillance due to high malignancy risk.

Case 4

A 14-year-old boy presented with recurrent abdominal discomfort and altered bowel habits for 1 year. Symptoms were mild but progressive. There was no history of bleeding or weight loss. On examination, mucocutaneous pigmentation was noted on lips and oral mucosa. Colonoscopy revealed multiple pedunculated polyps in the colon, the largest measuring 3 cm. Endoscopic polypectomy was performed. Histopathology confirmed hamartomatous nature of the polyps. Family screening revealed similar findings in his mother, confirming familial Peutz-Jeghers Syndrome. The patient was enrolled in a structured surveillance program and remained stable without complications at 3-year follow-up.

Case 5

A 30-year-old woman presented with progressive abdominal pain, unexplained weight loss, and occasional vomiting over 6 months. Clinical examination revealed cachexia and mucocutaneous pigmentation. Imaging studies showed multiple polyps along with a suspicious mass in the pancreas. Endoscopic biopsy confirmed early-stage pancreatic cancer associated with Peutz-Jeghers Syndrome. The patient underwent surgical resection followed by oncological treatment. This case highlighted the malignant potential of the syndrome. The patient is currently under close multidisciplinary follow-up. The clinical presentation, diagnostic findings, management, and outcomes of all cases are summarized in Table 1. "The characteristic clinical and radiological features of Peutz-Jeghers Syndrome, including mucocutaneous pigmentation, hamartomatous polyps, and intussusception, are illustrated in Figure 1."

DISCUSSION

Peutz-Jeghers Syndrome (PJS) is a rare autosomal dominant disorder characterized by hamartomatous gastrointestinal polyps and distinctive mucocutaneous pigmentation. First described by Jan Peutz in 1921 and later defined as a clinical entity by Harold Jeghers in 1949, the syndrome has since been extensively studied in terms of its genetic, clinical, and oncological implications (Beggs *et al*., 2010).

The global incidence of PJS is estimated to range from 1 in 50,000 to 1 in 200,000 individuals, affecting both genders equally. Approximately 50-60% of cases are familial, while the remaining arise due to *de novo* mutations. The condition remains underdiagnosed in developing regions due to limited access to genetic testing and lack of awareness (Matsubara *et al*., 2025).

At the molecular level, PJS is primarily caused by mutations in the STK11 (LKB1) gene, which encodes a serine/threonine kinase involved in regulating cell proliferation, polarity, and apoptosis. Disruption of the AMP-Activated Protein Kinase (AMPK) pathway leads to uncontrolled cellular growth and contributes to the development of hamartomatous polyps as well as increased cancer susceptibility (McGarrity and Amos, 2006; Klimkowski *et al*., 2021).

Pathophysiologically, the hallmark feature of PJS is the formation of hamartomatous polyps, predominantly in the small intestine, particularly the jejunum. These polyps can act as lead points for intussusception, resulting in bowel obstruction. Chronic bleeding from these lesions may also lead to iron deficiency anemia. The premalignant nature of PJS is well established, with cumulative lifetime cancer risk reported to be as high as 80-90%, involving

organs such as the colon, pancreas, stomach, breast, and ovaries (Klimkowski *et al.*, 2021; Giardiello and Trimbath, 2006).

Clinically, patients commonly present with abdominal pain, gastrointestinal bleeding, anemia, and characteristic mucocutaneous pigmentation involving the lips, oral mucosa, and extremities. These pigmented lesions often appear in early childhood and serve as important diagnostic clues, although they may fade with age. Complications such as intussusception and intestinal obstruction are frequently reported and may require urgent surgical intervention (Amru and Dhok, 2024).

Diagnosis of PJS is based on a combination of clinical findings, endoscopic evaluation, imaging studies, and genetic testing. Endoscopic techniques, including colonoscopy and enteroscopy, are essential for detecting and managing polyps, while imaging modalities such as CT and MRI aid in identifying complications. Genetic testing for *STK11* mutations remains the gold standard for confirmation (Kopacova *et al.*, 2009).

Management of PJS focuses on symptom control, prevention of complications, and surveillance for malignancy. Endoscopic polypectomy is the primary treatment for accessible polyps, whereas surgical intervention is indicated in cases of obstruction or intussusception. Long-term surveillance using endoscopy and imaging is crucial for early detection of malignancies. A multidisciplinary approach involving gastroenterologists, surgeons, and genetic counselors is essential for optimal patient care (Giardiello and Trimbath, 2006; Beggs *et al.*, 2010).

The illustrative cases presented in this study reflect the diverse clinical spectrum of PJS. Common features observed across cases included abdominal pain, anemia, and mucocutaneous pigmentation, consistent with previously reported findings (Pandit *et al.*, 2024; Amru and Dhok, 2024). Two patients presented with intussusception requiring surgical management, aligning with literature that identifies it as a major complication due to polyp-related lead points (Klimkowski *et al.*, 2021).

Endoscopic polypectomy was effective in managing accessible lesions in several cases, while surgical intervention was necessary in acute presentations. Histopathological confirmation of hamartomatous polyps was consistent across all cases, supporting its role as a diagnostic hallmark (Kopacova *et al.*, 2009).

Notably, one case demonstrated pancreatic malignancy, reinforcing the significantly elevated cancer risk associated with PJS. This observation is consistent with existing literature emphasizing the importance of long-term surveillance and early

Table 1: Summary of Clinical Presentation, Diagnostic Findings, Management, and Outcomes in Illustrative Cases of Peutz-Jeghers Syndrome.

Case	Age	Gender	Presentation	Findings	Management	Outcome
1.	12	Female	Abdominal pain, vomiting	Jejunal intussusception	Resection	Stable (5 yrs)
2.	19	Male	Melena, anemia	Multiple GI polyps	Polypectomy	Stable (4 yrs)
3.	25	Female	Acute obstruction	Ileal intussusception	Surgery	Recovered
4.	14	Male	Mild abdominal pain	Colonic polyps	Endoscopic removal	Stable
5.	30	Female	Weight loss, pain	Polyps + pancreatic cancer	Surgery + oncology	Under follow-up

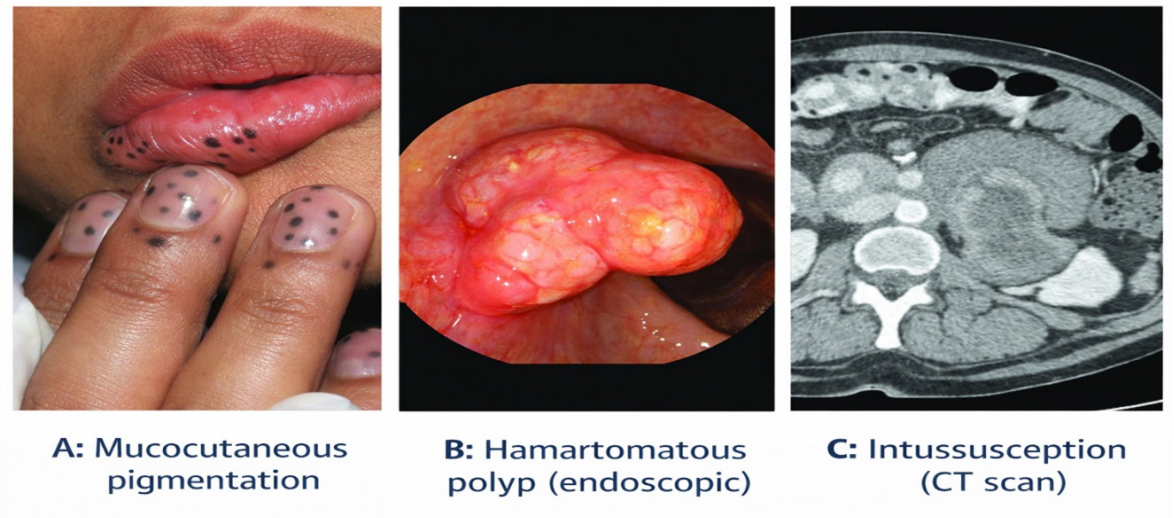

Figure 1: Key manifestations of Peutz-Jeghers Syndrome.

cancer detection in improving patient outcomes (Giardiello and Trimbath, 2006; Matsubara *et al*., 2025).

Overall, these findings highlight the importance of early recognition, appropriate intervention, and lifelong surveillance in improving clinical outcomes and reducing morbidity and mortality associated with Peutz-Jeghers Syndrome.

CONCLUSION

Peutz-Jeghers Syndrome is a rare but clinically significant disorder with varied presentations and potential for serious complications, including malignancy. Recognition of characteristic features, particularly mucocutaneous pigmentation, is essential for early diagnosis. Timely intervention and regular surveillance play a vital role in reducing morbidity and improving long-term prognosis.

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ABBREVIATIONS

AMPK: AMP-activated protein kinase; **CECT:** Contrast-enhanced computed tomography; **CT:** Computed tomography; **GI:** Gastrointestinal; **MRI:** Magnetic resonance imaging; **PJS:** Peutz-Jeghers Syndrome.

CONFLICT OF INTEREST

The authors declare that there is no conflict of interest.

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AUTHOR CONTRIBUTIONS

GH conceptualized the study. GP and PMM collected the clinical data. SM and KL drafted the manuscript. GRNG critically reviewed and edited the manuscript. All authors approved the final version.

SUMMARY

Peutz-Jeghers Syndrome is a rare genetic disorder requiring a high index of clinical suspicion, largely driven by the observation of mucocutaneous pigmentation and recurrent abdominal pain. As highlighted by the literature review and five diverse clinical cases, PJS poses severe risks such as intussusception and a highly elevated lifetime risk of various malignancies. The manuscript stresses that a multidisciplinary approach—combining endoscopic management, surgical intervention when necessary, and lifelong genetic and oncological surveillance—is essential for optimizing patient outcomes.

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